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April 9, 2010

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Publication List

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Pub Date	Source Identifier *(embargoed until)	PA Compliance	Pub Title	Journal (Impact)	Pub Authors
2010 Apr 5	PMID19908235 - -	Questionable Compliance	Psychological symptoms correlate with reduced hippocampal volume in fragile X premutation carriers.	American journal of medical genetics. Part B, Neuropsychiatric genetics : the official publication of the International Society of Psychiatric Genetics	Adams, P E; Adams, J S; Nguyen, D V; H Brunberg, J A; Tassone, F; Zhang, W; Ko K; Rivera, S M; Grigsby, J; Zhang, L; Dec Hagerman, P J; Hagerman, R J
2010 Jan 19	PMID20042704 PMCID2809036 -	PA Compliant	Alzheimer's Disease Neuroimaging Initiative (ADNI): clinical characterization.	Neurology	Petersen, R C; Aisen, P S; Beckett, L A; D M C; Gamst, A C; Harvey, D J; Jack Jr, C Jagust, W J; Shaw, L M; Toga, A W; Troja Q; Weiner, M W
2009 Dec 15	PMID20007524 PMCID2790222 -	PA Compliant	Association of parental dementia with cognitive and brain MRI measures in middle-aged adults.	Neurology (7.043)	Debette, S; Wolf, P A; Beiser, A; Au, R; Hi Pikula, A; Auerbach, S; Decarli, C; Sessa
2009 Dec	PMID19726595 PMCID2777470 -	PA Compliant	Metabolic evidence of vitamin B-12 deficiency, including high homocysteine and methylmalonic acid and low holotranscobalamin, is more pronounced in older adults with elevated plasma folate.	The American journal of clinical nutrition (6.74)	Miller, Joshua W; Garrod, Marjorie G; All Lindsay H; Haan, Mary N; Green, Ralph
2009 Nov	PMID19901172 - -	Questionable Compliance	Differences in brain volume, hippocampal volume, cerebrovascular risk factors, and apolipoprotein E4 among mild cognitive impairment subtypes.	Archives of neurology	He, Jing; Farias, Sarah; Martinez, Oliver; Bruce; Mungas, Dan; Decarli, Charles
2009 Nov 24	PMID19846830 PMCID2788808 -	PA Compliant	Regional pattern of white matter microstructural changes in normal aging, MCI, and AD.	Neurology (7.043)	Lee, D Y; Fletcher, E; Martinez, O; Ortega Zozulya, N; Kim, J; Tran, J; Buonocore, M Carmichael, O; DeCarli, C
2009 Sep	PMID19752306 - -	Questionable Compliance	Progression of mild cognitive impairment to dementia in clinic- vs community-based cohorts.	Archives of neurology	Farias, Sarah Tomaszewski; Mungas, D Bruce R; Harvey, Danielle; DeCarli, Char
2009 Jun	PMID19403891 PMCID2774231 NIHMS136203	PA Compliant	Do tests of executive functioning predict ability to downregulate emotions spontaneously and when instructed to suppress?	Cognitive, affective & behavioral neuroscience (3.132)	Gyurak, Anett; Goodkind, Madeleine S; M Anita; Kramer, Joel H; Miller, Bruce L; Le Robert W
	PMID19437501	Questionable Compliance	Cognitive decline in mild cognitive impairment		Xie, Jing; Alcantara, Dan; Amenta, Nina;



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[The emotional brain: combining insights from patients and basic science.](#)

1. Rosen HJ, Levenson RW.
 Neurocase. 2009 Jun;15(3):173-81.
 PMID: 20183547 [PubMed - in process]
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[Common variants at 7p21 are associated with frontotemporal lobar degeneration with TDP-43 inclusions.](#)

2. Van Deerlin VM, Sleiman PM, Martinez-Lage M, Chen-Plotkin A, Wang LS, Graff-Radford NR, Dickson DW, Rademakers R, Boeve BF, Grossman P, van Swieten JC, Murrell JR, Ghetti B, Spina S, Grafman J, Hodges J, Spillantini MG, Gilman S, Lieberman AP, Kaye JA, Woltjer RL, Bigio EH, Me Ferrer I, Lladó A, Neumann M, Kretzschmar HA, Hulette CM, Welsh-Bohmer KA, Miller BL, Alzualde A, de Munain AL, McKee AC, Gearing M, Levey Feldman HH, Hamilton RL, Dekosky ST, van der Zee J, Kumar-Singh S, Van Broeckhoven C, Mayeux R, Vonsattel JP, Troncoso JC, Kriol JJ, Kwok J, McLean CA, DeCarli C, Ellis WG, Freeman SH, Frosch MP, Growdon JH, Perl DP, Sano M, Bennett DA, Schneider JA, Beach TG, Reiman EM, Wojcicki I, Hartikainen P, Seilhean D, Galasko D, Masliah E, Cotman CW, Tuñón MT, Martínez MC, Munoz DG, Carroll SL, Marson D, Riederer PF, Bogdan VM.
 Nat Genet. 2010 Mar;42(3):234-9. Epub 2010 Feb 14.
 PMID: 20154673 [PubMed - in process]
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[Mapping Alzheimer's disease progression in 1309 MRI scans: power estimates for different inter-scan intervals.](#)

3. Hua X, Lee S, Hibar DP, Yanovsky I, Leow AD, Toga AW, Jack CR Jr, Bernstein MA, Reiman EM, Harvey DJ, Kornak J, Schuff N, Alexander GE, W Initiative.
 Neuroimage. 2010 May 15;51(1):63-75. Epub 2010 Feb 6.
 PMID: 20139010 [PubMed - in process]
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Common variants at 7p21 are associated with frontotemporal lobar degeneration with TDP-43 inclusions.

Van Deerlin VM, Sleiman PM, Martinez-Lage M, Chen-Plotkin A, Wang LS, Graff-Radford NR, Dickson DW, Rademakers R, Boeve BF, Grossman M, Arnold SE, Mann DM, Pickering-Brown SM, Seelaar H, Heutink P, van Swieten JC, Murrell JR, Ghetti B, Spina S, Grafman J, Hodges J, Spillantini MG, Gilman S, Lieberman AP, Kaye JA, Woltjer RL, Bigio EH, Mesulam M, Al-Sarraj S, Troakes C, Rosenberg RN, White CL 3rd, Ferrer I, Lladó A, Neumann M, Kretschmar HA, Hulette CM, Welsh-Bohmer KA, Miller BL, Alzualde A, de Munain AL, McKee AC, Gearing M, Levey AI, Lah JJ, Hardy J, Rohrer JD, Lashley T, Mackenzie IR, Feldman HH, Hamilton RL, Dekosky ST, van der Zee J, Kumar-Singh S, Van Broeckhoven C, Mayeux R, Vonsattel JP, Troncoso JC, Kril JJ, Kwok JB, Halliday GM, Bird TD, Ince PG, Shaw PJ, Cairns NJ, Morris JC, McLean CA, DeCarli C, Ellis WG, Freeman SH, Frosch MP, Crowdon JH, Perl DP, Sano M, Bennett DA, Schneider JA, Beach TG, Reiman EM, Woodruff BK, Cummings J, Vinters HV, Miller CA, Chui HC, Alafuzoff I, Hartikainen P, Seilhean D, Galasko D, Masliah E, Cotman CW, Tuñón MT, Martínez MC, Muñoz DG, Carroll SL, Marson D, Riederer PF, Bogdanovic N, Schellenberg GD, Hakonarson H, Trojanowski JQ, Lee VM.

[1] Department of Pathology and Laboratory Medicine, University of Pennsylvania School of Medicine, Philadelphia, Pennsylvania, USA. [2] These authors contributed equally to this work.

Frontotemporal lobar degeneration (FTLD) is the second most common cause of presenile dementia. The predominant neuropathology is FTLD with TAR DNA-binding protein (TDP-43) inclusions (FTLD-TDP). FTLD-TDP is frequently familial, resulting from mutations in GRN (which encodes progranulin). We assembled an international collaboration to identify susceptibility loci for FTLD-TDP through a genome-wide association study of 515 individuals with FTLD-TDP. We found that FTLD-TDP associates with multiple SNPs mapping to a single linkage disequilibrium block on 7p21 that contains TMEM106B. Three SNPs retained genome-wide significance following Bonferroni correction (top SNP rs1990622, $P = 1.08 \times 10^{-11}$); odds ratio, minor allele (C) 0.61, 95% CI 0.53-0.71). The association replicated in 89 FTLD-TDP cases (rs1990622; $P = 2 \times 10^{-4}$). TMEM106B variants may confer risk of FTLD-TDP by increasing TMEM106B expression. TMEM106B variants also contribute to genetic risk for FTLD-TDP in individuals with mutations in GRN. Our data implicate variants in TMEM106B as a strong risk factor for FTLD-TDP, suggesting an underlying pathogenic mechanism.

PMD: 20154673 [PubMed - in process]

PMCID: PMC2828525 [Available on 2010/9/1]

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Am J Med Genet B Neuropsychiatr Genet. 2010 Apr 5;153B(3):775-85.

Psychological symptoms correlate with reduced hippocampal volume in fragile X premutation carriers.

Adams PE, Adams JS, Nguyen DV, Hessl D, Brunberg JA, Tassone F, Zhang W, Koldewyn K, Rivera SM, Grigsby J, Zhang L, Decarli C, Hagerman PJ, Hagerman RJ.

M.I.N.D. Institute, University of California, Davis Health System, Sacramento, California, USA.

Fragile X-associated tremor/ataxia syndrome (FXTAS) is a neurodegenerative disorder occurring in male and occasional female carriers of a premutation expansion (55-200 CGG repeats) of the fragile X mental retardation 1 gene. We assessed the relationship between hippocampal volume and psychological symptoms in carriers, both with and without FXTAS, and controls. Volumetric MRI measures, clinical staging, cognitive testing, molecular analysis, and psychological symptoms were performed for female premutation carriers both with FXTAS (n = 16, age: 57.50 + or - 12.46) and without FXTAS (n = 17, age: 44.94 + or - 11.23), in genetically normal female controls (n = 8, age: 50.63 + or - 11.23), carriers with FXTAS (n = 34, age: 66.44 + or - 6.77) and without FXTAS (n = 21, age: 52.38 + or - 12.11), and genetically normal male controls (n = 30, age: 57.20 + or - 14.12). We examined the relationship between psychological symptoms and hippocampal volume, as well as correlations with molecular data. We found a significant negative correlation between total hippocampal volume and anxiety in female carriers, with and without FXTAS. This finding was mainly driven by a negative correlation between right hippocampal volume and anxiety. Other anxiety-related subscales also correlated with the right hippocampus in females. In male carriers with and without FXTAS, only paranoid ideation negatively correlated with hippocampal volume. Female premutation carriers demonstrated a negative association between hippocampal volume and the severity of anxiety-related psychological symptoms. Though the presentation of FXTAS symptoms is primarily in males, anxiety-related problems are common both prior to and after the onset of FXTAS, and may be related to hippocampal changes.

PMID: 19908235 [PubMed - in process]

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